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MEETING REPORT

Rare diseases as a global priority: the CIS Orphan Forum and the path toward collaboration with BRICS

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ABSTRACT

The Second Orphan Forum of the Commonwealth of Independent States, held in Moscow on June 26–27, 2025, served as a platform to translate World Health Assembly Resolution 78.11 on rare diseases into coordinated policy action. The Forum convened delegates from eight Commonwealth of Independent States members and representatives from India, the United Arab Emirates, South Africa, and Oman, advancing interregional cooperation.

Participants highlighted shared challenges: lack of national strategies and harmonized definitions of rare diseases, gaps in diagnostics and care infrastructure, limited registries, shortages of trained specialists, and unstable funding. The adopted Resolution set four priority domains for joint work: policy and regulatory development; organization of care

and workforce capacity; pharmaceutical provision and health technology assessment; and international collaboration.
Country presentations showed progress alongside persistent gaps. Priorities include expanding screening, establishing centers of expertise, improving patient pathways, extending reimbursement, introducing accelerated regulatory procedures, and updating clinical guidelines. Recommendations emphasize integrated care, continuity from pediatric to adult services, stronger health technology assessment for orphan medicines, real-world data collection, and managed entry and risk-sharing agreements.
The Forum also concluded that closer cooperation on rare diseases is needed within BRICS, especially to improve the efficiency of orphan medicine development, manufacturing, and procurement through coordinated approaches, demand aggregation, and joint price negotiations.

Key Words: orphan diseases, CIS Orphan Consortium, reimbursement policy, joint procurement, orphan medicines, regulatory harmonization

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Introduction

Rare diseases comprise a heterogeneous group of over 6,000 conditions, most of which are genetic in origin and manifest during childhood. Although each disease is rare (on average 1 case per 2,000 people), collectively they affect between 3.5 and 5.9 percent of the world's population (more than 450 million individuals) [1]. Effective therapies exist for fewer than 5% of these conditions, while the annual cost of treatment often exceeds 300,000–400,000 US dollars (USD) per patient [2]. Diagnostic delays, insufficient clinical expertise, and lack of integrated social support make rare diseases not only a medical but also a socioeconomic challenge.

In May 2025[1], the World Health Assembly (WHA) adopted Resolution WHA 78.11, officially elevating rare diseases to a global health priority. The Resolution emphasized the need to integrate rare diseases into national health strategies, expand screening programs, establish centers of excellence, national and regional registries, and innovative ways of funding. It lays the foundation for the ten-year World Health Organization (WHO) Global Action Plan on Rare Diseases, aimed at embedding rare diseases policy within Universal Health Coverage.

For the Commonwealth of Independent States (CIS) region, these priorities are of particular relevance. Despite growing interest, most states still lack comprehensive national strategies, legal definitions, or long-term funding models [3]. In 2024, the CIS Orphan Consortium was

[1] World Health Organization. Rare diseases: A global health priority for equity and inclusion. Accessed 11.12.2025. https://apps.who.int/gb/ebwha/pdf_files/WHA78/A78_R11-en.pdf

established as a multi-country platform bringing together experts, patient organizations, and regulators to coordinate policies and act as the WHO's regional partner in shaping the WHO Global Action Plan.

The II CIS Orphan Forum (Moscow, June 26–27, 2025) became the first regional event focused on implementing the WHA Resolution and strengthening collaboration with BRICS. Delegations from eight CIS countries (Republic of Azerbaijan, Republic of Armenia, Republic of Belarus, Republic of Kazakhstan, Kyrgyz Republic, Russian Federation, Republic of Tajikistan, Republic of Uzbekistan) participated alongside partners from India, the United Arab Emirates (UAE), South Africa, and Oman, creating a CIS–BRICS interregional dimension.

Opening the Forum, Ulrike Schwerdtfeger (Figure 1), Technical Lead for Rare Diseases at the WHO, presented the key priorities of Resolution WHA 78.11, the first-ever resolution on rare diseases discussed and adopted by the WHA.

Among the matters of high importance for WHO Member States as reflected in the resolution are the need for integration of rare diseases into national health systems including in national health policies and programmes; the urgent need for cross-sector collaboration to foster innovation in research and innovative diagnosis and treatment; the establishment of national, regional and international centers of excellence as specialized hubs for care, research and training for rare diseases, human resource capacity; the use of interoperable codification systems for rare diseases; the establishment of national and regional registries; patient engagement mechanisms and the adoption of innovative ways of funding and resource mobilization.

She also announced that WHO, in consultation with its Member States and other stakeholders, including patient organizations, will develop a draft 10-Year WHO Global Action Plan on Rare Diseases.

FIG. 1. Ulrike Schwerdtfeger, Technical Lead Rare Diseases at the WHO, presenting online

Note: Alexander Rumyantsev, President of CIS Orphan Consortium, President of the Federal State Budgetary Institution "Dmitry Rogachev National Medical Research Center for Pediatric Hematology and Oncology" of the Ministry of Health of the Russian Federation, Academician of the Russian Academy of Sciences and Elena Shamal, Advisor to the Department for Cooperation in Political, Humanitarian and Social Spheres of the CIS Executive Committee, Secretary of the Council for Cooperation in Healthcare of the CIS co-chairing the plenary session.

In preparation for the development of the Global Action Plan, WHO will map existing WHO standards, guidelines and protocols relating to rare diseases, and identify technological innovation opportunities (including e-health, m-health, digital and artificial intelligence solutions) to centralize clinical health information for diagnostics and treatment. To support improved access to diagnosis and care, WHO will also identify centers of excellence around the world able to cluster clinical work in rare disease groups and to act as hubs to exchange experience and clinical knowledge and provide peer-to-peer medical advice, including across borders[2].

Thus, the Forum became a pivotal mechanism for translating WHO's global principles into regional practice. Its agenda, ranging from national policy and regulatory frameworks to health technology assessment (HTA), real-world data (RWD)/real-world evidence (RWE), and financing, defined the contours of the future rare disease care ecosystem in the CIS and BRICS regions as an integral part of implementing Resolution WHA 78.11.

Plenary session I. Rare diseases in the focus of national strategy: coordination of efforts and development priorities

The first plenary session of the II CIS Orphan Forum opened a central discussion on the practical implementation of the WHA Resolution WHA 78.11. The session gathered representatives of the World Health Organization, ministries of health of CIS member states, the CIS Executive Committee, and international partners.

In the welcoming address, the Minister of Health of the Russian Federation, Mikhail Murashko, emphasized that the adoption of the WHA Resolution marks a new stage in consolidating global responsibility for rare diseases and reinforces the moral and political commitment of national health systems to ensure equal access to care for all patients.

Academician Alexander Rumyantsev, President of the CIS Orphan Consortium, recalled that the first Forum in 2024 initiated the creation of the Consortium, which now unites more than 20 leading medical centers and patient organizations. He stressed that the 2025 Forum had evolved into an interregional platform where the CIS and BRICS act as a unified area of shared responsibility for patients with rare diseases.

Delegates from CIS member states presented reports on the current status of medical care for patients with rare diseases. Kazakhstan demonstrated the country's transition from fragmented measures to a systemic model: 56 clinical protocols for diagnosis and treatment have been approved, national screening programs implemented, and a Republican Coordination Center and national patient registry established.

The Russian Federation's report underlined that Russia is building a comprehensive, multi-level system of care for rare disease patients from neonatal screening and federal expert centers to centralized pharmaceutical supply and the "Circle of Kindness" Foundation, which ensures equitable access to therapy for all children in the country.

[2] Schwerdtfeger U. World Health Assembly Resolution WHA78.11: Rare Diseases – A Priority for Global Health Equity and Inclusion [Online presentation]. II Forum of CIS Countries on Orphan Diseases. CIS Orphan Consortium. Accessed 11.12.2025. https://en.orphan-cis.net/events/92/

The representative of Uzbekistan highlighted the importance of multidisciplinary teams and integration with social services, while Kyrgyzstan emphasized the need to legally define the national list of rare diseases and establish its own regulatory framework in this field.

Plenary session II. The future of rare disease therapy: new technologies, accessibility, and partnership across the CIS, BRICS, and the Middle East

The second plenary session became the central international segment of the Forum and the first platform where the rare disease agenda was discussed within the context of the global technological and economic trends of the BRICS countries. The session focused on developing a joint sustainable model of access to innovative therapies, using instruments such as HTA, RWD/RWE, multi-criteria decision analysis (MCDA), centralized price negotiations and procurement, and mechanisms for collaborative research.

India

Prasanna Kumar Shirol (Figure 2), Co-Founder and Executive Director of the Organization for Rare Diseases India (ORDI), presented India's evolving experience in shaping a comprehensive national policy for rare diseases and the potential for extending this framework through cooperation within BRICS. Drawing on his dual perspective as both an advocate and a parent of India's first Pompe disease patient, Shirol described how patient-led activism in India has become a catalyst for policy reform, infrastructure development, and public awareness.

He outlined the main pillars of India's National Policy for Rare Diseases (2021), which established a network of Centres of Excellence, created a national registry under the Indian Council of Medical Research, and introduced state-supported treatment for 63 prioritized conditions. This policy, together with judicial advocacy and parliamentary oversight, has ensured a gradual increase in national funding for rare diseases – from pilot grants to a sustained budget line under the Ministry of Health. These developments have been described in prior policy analyses [4, 5]. The Production-Linked Incentive scheme and customs duty exemptions have further encouraged domestic production of orphan drugs and medical devices, reducing dependency on imports and aligning industrial incentives with patient needs.

Shirol emphasized the importance of integrating research, innovation, and access, noting recent initiatives such as the National Consortium for Research and Development on therapeutics for Rare Diseases and a newly announced 1 million USD government prize for the development of a drug for sickle-cell disease. He stressed that India's combination of robust pharmaceutical manufacturing, digital health infrastructure, and active civil society provides a scalable foundation for cross-border collaboration.

In his strategic proposal titled "Rare BRICS: A Collaborative Framework for Rare Diseases," Shirol called for the establishment of a BRICS-wide alliance bringing together regulators, clinicians, researchers, and patient organizations. The initiative would aim to:

- harmonize definitions and orphan-drug designations;
- develop a joint research and manufacturing consortium to focus on priority conditions;

FIG. 2. Prasanna Kumar Shirol, Co-Founder and Executive Director of the Organization for Rare Diseases India

- coordinate Good Clinical Practice standards and facilitate fast-track approval of orphan drugs recognized by any BRICS regulatory agency;
- explore joint procurement and price-negotiation models using existing mechanisms such as Russia's "Circle of Kindness" as templates for sustainable financing;
- advance telehealth, artificial intelligence-based genetic diagnostics, and cross-border training programs to build shared capacity.

Shirol concluded by underscoring that with 40% of the world's population residing in BRICS countries, the bloc has the demographic scale and technological capacity to redefine access to orphan therapies. Coordinated action among BRICS members, he argued, could transform the region into a global innovation hub for rare disease research, development, and equitable care[3].

United Arab Emirates

Dr. Ayman El-Hattab (Figure 3), consultant in clinical genetics and director of the Genetics and Rare Disease Center at Burjeel Medical City (Abu Dhabi), professor at the University of Sharjah

[3] Shirol P. K. BRICS – Way to collaborate. [Online presentation]. II Forum of CIS Countries on Orphan Diseases. CIS Orphan Consortium. Accessed 11.12.2025. https://en.orphan-cis.net/events/92/

and president of the Middle East and North Africa (MENA) Congress for Rare Diseases, outlined the broader context of rare diseases across the MENA region. Although the region is home to more than 600 million people across 24 countries, rare diseases remain under-recognized in public health policy. The majority of conditions are genetic, largely due to high consanguinity rates and large family size, yet registries are scarce, and diagnostic delays are common. Competing health priorities and limited data on prevalence and costs continue to hinder policy development [6, 7].

Focusing on the UAE, Dr. El-Hattab emphasized that the country has emerged as a regional leader in rare-disease policy and an active participant in the evolving BRICS health agenda. The UAE has introduced accelerated orphan-drug registration pathways based on European Medicines Agency and Food and Drug Administration approvals and implemented flexible managed-entry agreements negotiated among the Ministry of Health, Abu Dhabi public healthcare provider SEHA and the Dubai Health Authority [8]. These contracts, mainly financial, with growing use of outcome-based models, address the high cost of orphan

FIG. 3. Dr. Ayman El-Hattab, consultant in clinical genetics and director of the Genetics and Rare Disease Center at Burjeel Medical City (Abu Dhabi), professor at the University of Sharjah and president of the Middle East and North Africa Congress for Rare Diseases

drugs, which remain several times more expensive than conventional therapies. Among key national achievements are the Emirati Genome Project, launched in 2019 to sequence one million citizens; the Genetics and Rare Disease Center at Burjeel Medical City, offering integrated clinical and research services; and the Abu Dhabi Rare Disease Registry, embedded in the Malaffi electronic-health-record system to link clinical and genomic data for policy and research use. Preliminary results from this program have been partially reported [9].

Dr. El-Hattab concluded that the UAE is now entering a phase of "sustainable access," prioritizing national guidelines for orphan-drug designation, cost-effectiveness thresholds for ultra-rare diseases, and joint procurement mechanisms across the region. As a new member of BRICS, the UAE can serve as a bridge between the Middle East, the CIS and other BRICS countries, facilitating alignment of regulatory standards, collaborative clinical trials and equitable access to orphan therapies[4].

South Africa

Dr. Helen Malherbe (Figure 4), Associate Professor at the Centre for Human Metabolomics at North-West University (Potchefstroom), presented South Africa's efforts towards developing a rare-disease framework and its potential role within the BRICS health agenda.

With a population of about 63 million, the country faces a dual burden of communicable and non-communicable diseases, while rare conditions remain largely unrecorded in health statistics. More than 90 percent of congenital disorders go underreported, leaving an estimated 4.2 million people affected by rare diseases alone, with the average diagnostic delay exceeding 5 years, if diagnosed at all [10]. Treatments are scarce and often unaffordable in the public sector. Although South Africa has a strong legislative foundation, including the National Health

FIG. 4. Online presentation by Dr. Helen Malherbe, Associate Professor at the Centre for Human Metabolomics at North-West University (Potchefstroom)

[4] El-Hattab A. W. Rare Diseases in the MENA region. [Online presentation]. II Forum of CIS Countries on Orphan Diseases. CIS Orphan Consortium. Accessed 11.12.2025. https://en.orphan-cis.net/events/92/

Act, the Medicines Act, and the National Health Insurance Act (2023), implementation of rare-disease policy remains fragmented.

Dr. Malherbe noted that the national capacity for medical genetics and counselling remains insufficient, but recent initiatives are beginning to address this gap [11]. Ongoing efforts include the drafting of a National Rare Diseases Framework, work by the established South African Rare Diseases Access Initiative, and active civil-society engagement through Rare Diseases South Africa. Among near-term priorities are completing a rare disease national strategy for implementation by 2027, creating a patient-initiated rare disease population registry and newborn-screening roadmap, extending access to genetic services across all provinces, and embedding rare-disease education into medical curricula.

In her concluding remarks, Dr. Malherbe underscored the importance of strengthening BRICS–CIS collaboration through context-specific approaches rather than importing European models. She proposed creating a joint BRICS–CIS registry platform, coordinated workforce training through virtual fellowships and hospital partnerships, and an open-access policy toolkit for shared strategic planning. South Africa, she argued, could serve as a pilot site for integrated rare-disease services in resource-limited contexts, drawing on its experience in congenital-disorder policy and universal-health-coverage reform. Aligning national actions with the forthcoming WHO Global Action Plan on Rare Diseases would allow South Africa to contribute regional expertise to global progress while advancing equitable access and capacity-building across the BRICS partnership[5].

The plenary discussions also featured contributions from the Sultanate of Oman, which, although not a BRICS member, plays an increasingly important role in linking the CIS and Gulf regions. Dr. Ahmed Al Saidi, former Minister of Health of Oman, outlined the country's healthcare transformation within the framework of Oman Vision 2040 [12]. He explained that the national system, while achieving significant progress in primary and preventive care, continues to face challenges typical for small and mid-income states: limited genetic-diagnostic capacity, high dependence on imported medicines, and the rising cost of treatment for chronic and rare conditions [13]. To address these issues, Oman is expanding regional partnerships in the pharmaceutical and biotechnology sectors, aiming to localize production of essential and high-cost drugs, attract investment, and strengthen quality-control and regulatory capacity. The Ministry of Health also promotes the creation of joint supply chains and pooled procurement initiatives with Gulf and Asian partners to improve affordability and ensure continuity of access. This approach is supported by Dubois et al. (2021), who analyzed pooled procurement procedures for medicines in low- and middle-income countries, including Oman [14]. Dr. Al Saidi emphasized that closer scientific and industrial cooperation between Oman, the CIS and BRICS countries could enhance efficiency, support technology transfer, and increase the sustainability of rare-disease care across the wider Eurasian and Middle Eastern region[6].

Overall, the plenary discussions showed that the CIS and BRICS countries face comparable challenges in organizing care and ensuring access to effective therapies for rare diseases. At the same time, participants proposed practical measures for closer cooperation, including the harmonization of drug registration requirements,

[5] Malherbe, H. Rare diseases in South Africa [Online presentation]. II Forum of CIS Countries on Orphan Diseases. CIS Orphan Consortium. Accessed 11.12.2025. https://en.orphan-cis.net/events/92/

[6] Al Saidi A. II Forum of CIS countries on Orphan Diseases. Transforming Healthcare, Innovations and Challenges In Modern Medicine. [Online presentation]. II Forum of CIS Countries on Orphan Diseases. CIS Orphan Consortium. Accessed 11.12.2025. https://en.orphan-cis.net/events/92/

the development of shared mechanisms for joint procurement, and the exchange of clinical knowledge and professional expertise. The Forum thus marked a transition from fragmented national actions to coordinated regional dialogue, outlining the foundations for a BRICS–CIS Alliance for Rare Diseases aimed at improving availability and equity of treatment across regions.

Nosological and thematic sessions: identified barriers and directions for solutions

The thematic and disease-specific sessions of the second day of the Forum (Figure 5) provided an in-depth analysis of how the challenges of organizing care and ensuring access for patients with rare diseases are reflected across different medical and institutional contexts. Despite diverse national systems, countries of the CIS region demonstrated a shared pattern of fragmentation, limited data, and gaps in continuity of care. The sessions also produced a number of common proposals that were subsequently incorporated into the Resolution of the II CIS Orphan Forum[7].

FIG. 5. Participants of Nosological and thematic sessions 2 days of the II CIS Orphan Forum

[7] Резолюция по итогам II Орфанного форума стран СНГ. Москва, Россия [CIS Orphan Consortium. Resolution of the II Orphan Forum of CIS Countries. Moscow, Russian Federation] (In Russian). Accessed 11.12.2025. https://orphan-cis.net/upload/iblock/ de6/7a5sp3cwz18lq67u6tb4py5r4xo8rudm/Rezolyutsiya_rus.pdf

Bleeding disorders

The session on bleeding disorders served as a model for identifying typical clinical and organizational challenges in rare-disease management. Experts from Russia, Kazakhstan, Tajikistan, Azerbaijan, and Kyrgyzstan emphasized the fragmented referral systems and shortage of specialized centers for hemophilia and other coagulopathies. Key problems include the absence of national registries, unequal access to medicines, and lack of continuity in adult care. Participants called for a transition from on-demand to preventive therapy, the establishment of multidisciplinary teams, and advanced training for hematologists, surgeons, and orthopedists to manage patients receiving modern therapies.

Rare tumors

The session on rare tumors brought together heads of pediatric oncology centers from Russia, Kazakhstan, Belarus, and Uzbekistan. Discussions focused on rare childhood cancers as one of the most complex segments of rare diseases. The main barriers identified were the absence of national registries, diagnostic errors reaching up to 20%, and insufficient laboratory capacity for molecular and morphological verification. Participants underlined the importance of establishing reference centers, developing biobanks, expanding the use of CAR-T therapies, and building interstate expert networks for the joint development of clinical recommendations.

Spinal muscular atrophy

The session on spinal muscular atrophy showcased successful examples of multisectoral collaboration. Russia reported neonatal screening coverage reaching 98 percent, while Kazakhstan and Uzbekistan presented national programs combining state and charitable funding [15]. Persistent problems include weak coordination during transition from pediatric to adult care, the absence of long-term reimbursement mechanisms, and limited registries for outcome monitoring. These challenges correspond to clauses 1.2, 1.4, and 1.12 of the Forum Resolution, which call for the institutionalization of centers, expansion of screening programs, and sustainable sources of financing.

Integration of care for rare diseases: multidisciplinarity and continuity at all stages of life

This session emphasized that rare diseases are inherently complex, often involving multiple organ systems and requiring the participation of specialists from different disciplines. Ensuring integration of care therefore entails several dimensions: interdisciplinary coordination among physicians and allied professionals, vertical integration between levels of care (from primary and specialized outpatient to inpatient services), and horizontal integration between regions and institutions to reduce territorial disparities.

Participants agreed that the most critical challenge lies in maintaining continuity of treatment during the transition from pediatric to adult care. Differences in drug-provision systems and organizational models of medical assistance frequently result in therapy interruptions, undermining the progress and investments achieved during childhood. In the past, few patients with rare diseases survived into adulthood, but earlier diagnosis and effective therapies have changed this trajectory. As the number of adult patients continues to grow, establishing integrated and continuous care pathways has become a top priority to preserve treatment outcomes and quality of life throughout the patient's lifespan.

Organization of the system of care for rare diseases: orphan centers, regional and national coordinators

Participants from Kazakhstan, Russia, Belarus, Uzbekistan, and Kyrgyzstan confirmed that most CIS countries still lack a formalized network of orphan centers and legally defined responsibilities for coordinators. Problems of patient routing, insufficient staffing, and weak links between medical, rehabilitation, and social services cause loss of continuity and exacerbate regional inequalities. The Resolution recommends establishing national and regional centers, securing their legal status and funding, incorporating rare-disease topics into medical education, and developing telemedicine cooperation among CIS countries.

Medicines reimbursement and health technology assessment

The session gathered experts from various CIS countries who discussed the use of HTA to support decision-making on innovative therapies for rare diseases. While HTA practices exist across the region, they are formally institutionalized only in Belarus, Kazakhstan, and Russia. Speakers pointed to the high level of uncertainty in clinical and economic evidence and the lack of RWD and RWE, particularly for gene and cell therapies, as key limitations. These gaps hinder robust evaluation and make conventional cost-effectiveness thresholds inadequate for rare diseases.

Participants highlighted the need to implement managed entry agreements as a way to combine early access with evidence generation. However, existing regulatory frameworks prevent most CIS countries from using such mechanisms. Belarus remains the only country with practical experience in managed entry agreements for high-cost orphan medicines, while others are exploring similar approaches but require legal reform. The session concluded that regional cooperation should focus on enabling performance-based reimbursement, improving data systems, and advancing toward joint clinical assessments within the Eurasian Economic Union (EAEU) and joint procurement initiatives across the CIS and BRICS frameworks to enhance affordability and access.

Pharmaceutical policy for rare diseases: challenges, approaches, perspectives

This session addressed the absence of harmonized definitions of rare diseases and registration procedures for orphan drugs across the EAEU. Experts noted that the lack of a unified recognition process leads to duplication of evaluations and delays in patient access to therapy. It was proposed to introduce a shared mechanism for granting orphan-drug status at the EAEU level and to include a dedicated chapter on rare diseases in the CIS Model Law on Pharmaceutical Provision.

Role of patient organizations and charitable foundations

The final session underlined the evolution of patient and charitable organizations from advocacy movements to recognized policy partners. Representatives from Russia, Kazakhstan, Kyrgyzstan, and Uzbekistan showed that patient associations are instrumental in monitoring therapy access and promoting expansion of screening programs. The Resolution calls for the institutional support of such organizations and for their involvement in planning and evaluating national rare-disease strategies.

It can be noted that patient organizations for rare diseases in the CIS countries are gaining a level of influence in specific decision-making processes comparable to that in Europe [16, 17].

In summary, the nosological and thematic sessions revealed a unified pattern of systemic challenges across the CIS region: the absence of comprehensive national strategies, legal definitions, and registries; a shortage of specialized personnel; fragmented financing; and weak inter-country coordination. These findings provide the foundation for the next section of the article, which analyzes the systemic barriers outlined in the Resolution of the II CIS Orphan Forum as a roadmap for strengthening regional collaboration on rare-disease policy.

Systemic problems identified following the II CIS Orphan Forum

An analysis of the Forum discussions and the adopted Resolution made it possible to systematize the main barriers hindering the development of effective rare-disease care systems in CIS countries. These challenges are grouped into four categories – regulatory and legal, organizational and infrastructural, pharmaceutical and economic, and analytical and methodological, in accordance with Section II of the Resolution.

Regulatory and legal problems

A clear and predictable regulatory and legal framework is the foundation of rare disease policy: it defines eligibility, establishes official lists and care pathways, and sets rules for funding, access, and accountability. Clear rules matter for every stakeholder: patients gain transparent and equitable access criteria, industry gains predictable requirements and planning horizons for launch and evidence generation, and health systems gain the ability to budget, negotiate, and manage resources more efficiently. In the CIS context, the Forum discussions highlighted several regulatory gaps that prevent rare disease care from developing into a sustainable system:

- Lack of national strategies and action plans for rare diseases. None of the countries in the region has yet approved a comprehensive strategy covering diagnosis, pharmaceutical provision, social support, and workforce development. Policies remain fragmented, decisions ad hoc and short-term, preventing systemic planning and evaluation.
- Inconsistency in definitions and lists of rare diseases. No unified definition or national lists exist. In Armenia, Azerbaijan, Kyrgyzstan, Tajikistan, and Uzbekistan, the regulatory framework is only being developed; in other countries, criteria differ, making statistical comparison difficult and hindering reimbursement-policy design.
- Non-harmonized procedures for determining orphan-drug status. Within the EAEU, orphan status is assigned at the national level, which delays registration and limits access to therapy. The lack of a single procedure reduces the efficiency of the common market and excludes some states from mutual recognition.
- Absence of sustainable financing mechanisms. Funding relies on regional or short-term programs without long-term commitments, creating inequity in access, particularly during transition from pediatric to adult care.
- Inability to implement flexible pharmaceutical-access models. There is no legal framework for managed-entry agreements, conditional reimbursement, or outcome-based payment. Post-marketing monitoring and review of funding decisions are not legislatively secured.

Organization of care and human resources

Rare disease care depends on coordinated service delivery: timely diagnosis, referral to specialized teams, long-term follow-up, and continuity across life stages. Because expertise is scarce and cases are complex, systems typically rely on defined networks of centers, standardized care pathways, and structured workforce development. The Forum discussions showed that in many CIS countries these elements remain informal or fragmented, which directly affects diagnostic delays, treatment continuity, and patient outcomes. The main points of the discussions included:

- Absence of an institutionalized network of orphan centers. In most countries, no legally recognized centers of competence with uniform standards, criteria, and financing exist. Care is delivered through isolated institutions without coordination or patient routing.
- Discontinuity between pediatric and adult services. Transitions occur unsystematically; mechanisms for communication and data transfer are lacking, leading to loss of follow-up and interruption of therapy.
- Workforce shortages and lack of training programs. Rare-disease topics are not included in medical curricula; no standardized postgraduate training or professional standards exist, limiting diagnostic and treatment quality.
- Insufficient cross-sectoral integration. Coordination between medical, social, educational, and rehabilitation services remains informal, and patient pathways often depend on individual initiative rather than institutional design.

Pharmaceutical access and health technology assessment

Access to orphan therapies requires balancing clinical need with financial sustainability under high uncertainty – small trials, limited comparators, and rapidly evolving evidence. HTA and related decision mechanisms help systems make transparent choices, structure price negotiations, and design adaptive access models (including monitoring and reassessment). The Forum discussions underlined that without institutional HTA capacity and real-world monitoring infrastructure, CIS countries struggle to implement consistent, value-oriented access policies for rare disease medicines. The raised concerns were as follows:

- High therapy costs and limited evidence base. Orphan drugs are expensive and supported by limited clinical data, complicating financing decisions in resource-constrained settings.
- Absence of institutionalized HTA structures. In Azerbaijan, Armenia, Kyrgyzstan, Tajikistan, and Uzbekistan, no official HTA agencies or units exist, preventing consistent evaluation and transparency.
- Methodological limitations of existing HTA systems. Even where HTA is operational (Russia, Kazakhstan, Belarus), methods are not adapted to orphan technologies.
- Lack of RWD/RWE systems. All CIS countries experience shortages of patient registries and monitoring mechanisms, making it impossible to review funding decisions dynamically or to use outcome-based models.
- Lack of HTA coordination across CIS countries. No unified methodologies or mutual recognition mechanisms exist, leading to duplicated work, higher costs, and reduced comparability.
- Underutilization of joint-procurement potential. Each country negotiates with manufacturers separately, losing economies of scale. The absence of consolidated negotiation platforms leads to higher prices.

Cross-country cooperation and data

For rare diseases, cross-country collaboration is not an "extra" but a practical necessity: patient populations are small, evidence generation requires pooling data, and negotiating power increases when countries coordinate. Shared registries, aligned regulatory/HTA approaches, and joint initiatives can reduce duplication, accelerate learning, and improve affordability. The Forum discussions highlighted that limited coordination and weak data ecosystems remain major constraints for both CIS and broader international cooperation. The particlural issues mentioned were:

- Lack of coordination and harmonization in regulation and HTA. Most CIS and BRICS countries still lack stable mechanisms for coordinating regulatory, assessment processes.
- Low participation in international research and registries. Countries in the region are underrepresented in global research networks, limiting access to advanced practices and international collaboration.
- Absence of joint initiatives for research and development (R&D) and local manufacturing. Despite existing industrial potential, there are no systemic projects for coordinated planning, registration, or production of orphan drugs.
- Insufficient use of joint-procurement opportunities. The lack of consolidated purchasing mechanisms within the CIS and BRICS reduces efficiency and limits therapy affordability.
- Deficit of reliable data and transparency. Many countries lack open patient registries and data on treatment outcomes, which hinders both management and international cooperation.

The combination of these problems demonstrates that CIS health systems are not yet fully ready to implement coordinated rare disease policy. To address these barriers, the Forum participants formulated a detailed set of recommendations – a regional "roadmap" for implementing the global rare-disease agenda presented in Section III of the Resolution.

Recommendations of the II CIS Orphan Forum

The outcomes of the II CIS Orphan Forum define a consistent framework for developing rare-disease policy across the CIS and BRICS regions. The recommendations cover four complementary dimensions: national policy and regulation, organization of care and human resources, analytical and methodological development, and international collaboration.

At the level of state policy, the Forum identified the adoption of national strategies and long-term action plans on rare diseases as a fundamental step. These documents should formalize priorities in diagnosis, screening, treatment, and rehabilitation, and provide a sustainable financial basis for patient care. Harmonization of definitions and criteria for rare diseases, as well as the establishment of a unified procedure for granting orphan-drug status within the EAEU, would improve coherence of regulation and accelerate patient access to therapy. The strengthening of legal and methodological frameworks is also necessary for the introduction of flexible reimbursement mechanisms, including managed entry agreements and risk-sharing models. Expanding neonatal and selective screening programs, and incorporating rare-disease topics into medical and postgraduate education, are integral elements of these reforms.

In the organization of care, the establishment of a structured network of national and regional orphan centers is identified as a key priority. These centers should serve as reference institutions responsible for patient routing, coordination of diagnostics and treatment, methodological guidance, and professional training. Their institutionalization will ensure uniform standards of care and improve access to expertise across the region.

Equally important is the need to strengthen continuity of care between pediatric and adult services. The absence of established transition mechanisms often leads to treatment interruption when patients move from children's to adult healthcare systems. To preserve therapeutic outcomes and ensure lifelong management, countries should develop unified protocols for transition, aligned drug-provision schemes, and coordination between inpatient, outpatient, and social-support services.

Systematic workforce development through training, internships, and coordinated educational programs across the CIS will help overcome existing human-resource shortages. Cross-border telemedicine and cooperation among orphan centers can ensure a more uniform level of expertise and continuity of care throughout the patient's life.

In the analytical and methodological dimension, the Forum emphasized the need to establish institutionalized HTA systems across the CIS countries, integrated into reimbursement and pricing decisions. The further development of HTA methodologies for orphan medicines should be informed by international experience and adapted to the specific features of rare diseases, with greater use of flexible evaluation criteria, differentiated willingness-to-pay thresholds, and patient-reported outcomes. Building national and regional registries and databases for RWD and RWE is essential to inform outcome-based models. The introduction of horizon scanning is required to anticipate emerging technologies and to plan for orphan-drug demand, enabling more sustainable and data-driven decision-making in healthcare budgeting and procurement. Centralization of procurement and the creation of joint price negotiations mechanisms could improve efficiency and ensure predictable pricing.

Finally, the Forum underscored the importance of international collaboration. Within the CIS, EAEU, and BRICS frameworks, coordinated demand forecasting, R&D coordination, shared data platforms, and joint price negotiations for orphan medicines would enhance sustainability and access.

The CIS Orphan Consortium is expected to continue serving as a key coordination platform for policy monitoring, comparative analysis, and the creation of an integrated information portal covering patients, centers, medicines, and treatment outcomes.

Taken together, these recommendations outline a regional roadmap for the formation of a sustainable and equitable system of care for patients with rare diseases. Their implementation will strengthen the emerging CIS–BRICS Alliance for Rare Diseases and contribute to achieving the goals of the WHO Global Action Plan on Rare Diseases.

Conclusion

The II CIS Orphan Forum confirmed that rare diseases have become an integral part of the global health agenda and a new area for consolidating the efforts of CIS and BRICS countries. The outcomes of the Forum and its adopted Resolution demonstrate the region's readiness to move from fragmented initiatives to a coordinated, long-term policy based on solidarity, knowledge exchange, and equitable access to innovative therapies.

The discussions and initiatives launched during the Forum laid the foundation for the creation of a CIS–BRICS Alliance for Rare Diseases – a multilateral platform uniting regulatory authorities, expert centers, and patient organizations to coordinate research, harmonize regulatory requirements, and promote joint R&D and access to orphan medicines. This Alliance is envisioned as a mechanism for sustained collaboration between CIS and BRICS countries.

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